The Economic and Social Burden of Spinal Muscular Atrophy (SMA) in the Italian Context

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Background

Spinal muscular atrophy (SMA), is a rare neuromuscular genetic disorder of the nervous system that occurs when the subject inherits from both parents mutations in the gene responsible for the disease. Patients with SMA experience a progressive worsening of their quality of life, which involves the impossibility to perform work activities or to perform them at full capacity, but in some cases the inability to perform simple daily activities. For this reason, most SMA patients need the assistance of a caregiver. SMA has 5 types, with varying degrees of severity: type 0, I, II, III, IV. The discovery of innovative treatments, especially in the case of rare diseases, is accompanied by economic needs and investment for the purchase of these therapies by the National Health Service (SSN). Nowadays, there are not enough economic information in the literature on the costs related to the disease, but it is possible to infer the considerable clinical and economic burden, aggravated by the high degree of morbidity to which patients with SMA are exposed. Recently, a German study has estimated that the average cost of the disease is 70,566 euros per year per patient.

Objectives

The purpose of this work is to assess, through a questionnaire provided to the association of patients (SMA Families) the economic and social burden of this genetic disease in Italy, developing a cost-of-illness model able to calculate the direct and indirect costs related to SMA in Italian context. The COI study represents a very useful tool in order to obtain a qualitative improvement of decisions in health care, especially when approached from a social perspective.

Methods

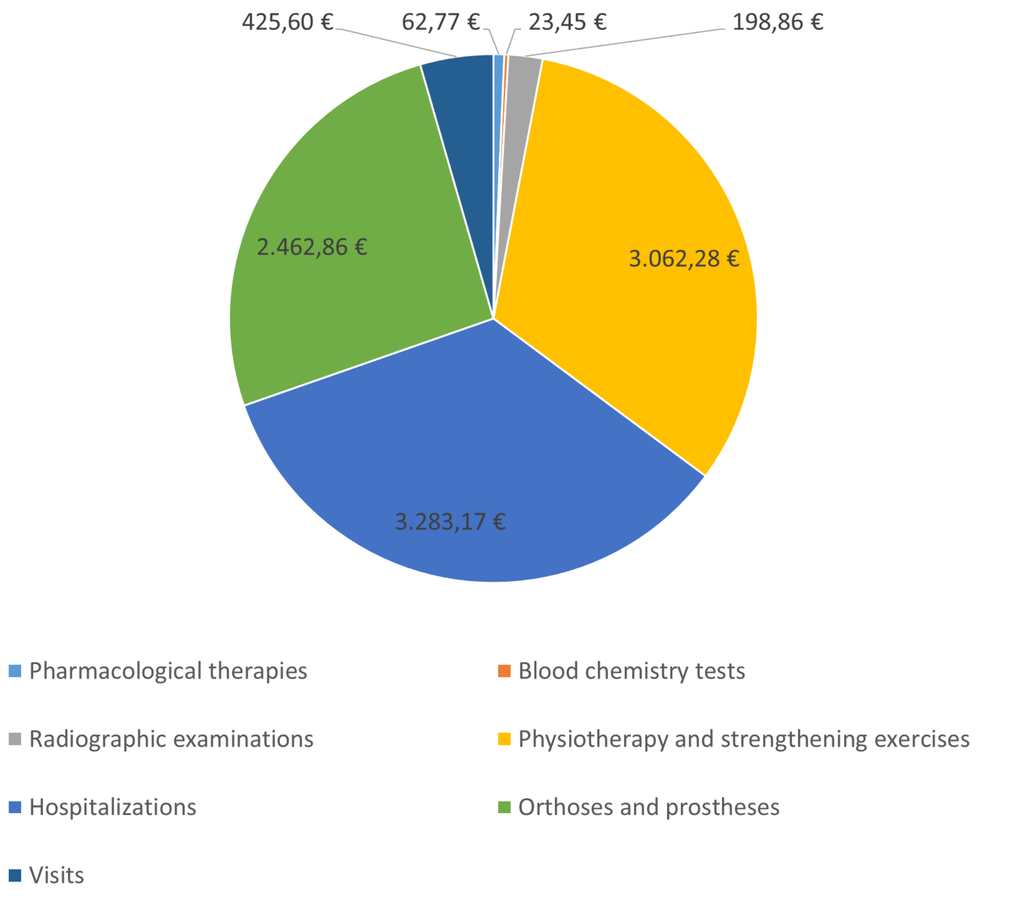
The cost-of-illness model represents an analysis of the costs incurred by society due to a specific disease. Both direct costs and productivity losses are included, thus including the point of view of society and not only that of the National Health Service. A questionnaire was developed and administered to experienced clinicians and to patient association in order to identify the uptake of resources directly imputable to the National Health Service (SSN), indirect costs (expressed in productivity losses for the patient and caregiver), and out-of-pocket expenses of patients and their families. In addition, to integrate the information obtained through the patients' association, some information has been completed or adapted using a questionnaire conducted by clinicians specialized in this pathology. For the purposes of the survey were collected data on 73 respondents, of which 53% were patient's caregiver with SMA. The model was developed through the method of activitity based costing (ABC), an useful tool for the determination of the resources absorption and the subsequent evaluation of the full cost of the interventions under analysis. The questionnaire is divided into 3 sections, personal data, pathology of the patient with SMA and direct non-healthcare costs.

Results

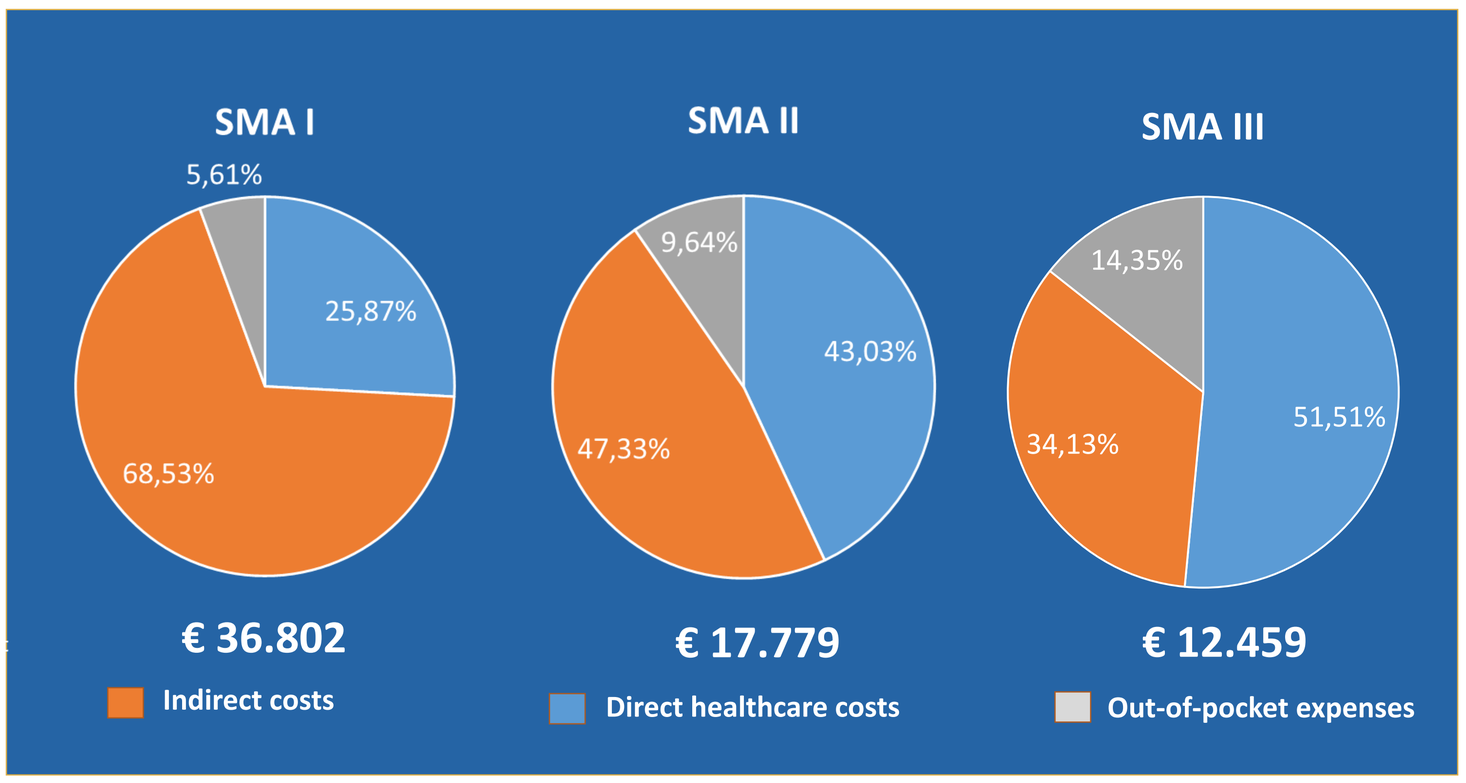
The results of the cost-of-illness analysis from the questionnaire analysis described above will be divided into: Direct costs, indirect costs, and out-of-pocket expenses.

- Direct healthcare costs are identified through the survey to patient associations and have the perspective of the National Health System. Thus, the treatments used by patients in the time horizon of the analysis, the analyses and consultations patients required, hospitalizations and specialist outpatient visits are included.
- Indirect costs are based on the social perspective, therefore, in order to determine the productivity losses of the patient and/or caregiver, expressed in terms of work days lost due to the condition, the 'human approach' is used. Therefore, the annual working hours that are normally carried out are considered and whether respondents were beneficiaries of the law 104/92, in order to make appropriate adjustments to the days of work lost and at the same time estimate the social impact of SMA.
- Out of pocket expenses are taken into consideration, i.e. those direct non-healthcare costs related to support teachers, the presence of a baby-sitter, transportation costs and days spent in a private facility. In this context, the visits carried out in private practice during the last year were also valued.

Taking into account the estimates reported in the previous paragraphs, the total cost of an SMA patient is equal to 19,884.47 € (excluding SMA specific therapies) considering the direct health costs (7,521.10 €) that represent 37.82% of total expenditure, the indirect costs (10,207.25 €) represent 51% of expenditure and the out-of-pocket expenses represent 10.84% of expenditure being valued at 2,156.10 €.



Direct medical costs	7.521,10 €
Indirect costs	10.207,28 €
Out of pocket expenses	2.156,10 €
SMA Therapies	62.534,25 €



Conclusions

The study showed an average annual cost for a patient with SMA in Italy of € 19,884.47 (excluding the costs of drug therapy specific for SMA). The study demonstrates an increasing cost as a function of the severity of the disease. Moreover, SMA is related to high social costs (indirect and out-of-pocket expenses). The purpose of this is to provide a valuable support for decision makers.